

WHAT IS CLAIMED IS:

- 1 1. An isolated nucleic acid comprising a sequence selected from the
2 group consisting of SEQ ID NO:9, SEQ ID NO: 10, SEQ ID NO: 11, and SEQ ID NO:12.
- 1 2. The isolated nucleic acid of Claim 1, wherein said nucleic acid is
2 deoxyribonucleic acid.
- 1 3. An isolated nucleic acid, wherein said nucleic acid is the complement
2 of the nucleic acid of Claim 1.
- 1 4. A vector comprising the nucleic acid of Claim 1.
- 1 5. A host cell comprising the vector of Claim 4.
- 1 6. An isolated protein encoded by the nucleic acid sequence set forth in
2 SEQ ID NO:9.
- 1 7. A method for detecting mutations in *Rab38* comprising the steps of:
2 a) amplifying at least a portion of *Rab38* from genomic DNA to
3 yield a *Rab38* amplification product;
4 b) purifying said *Rab38* amplification product; and
5 c) sequencing said *Rab38* amplification product.
- 1 8. The method of Claim 7, wherein said amplifying is accomplished
2 using a polymerase chain reaction.
- 1 9. The method of Claim 7, wherein said at least a portion of *Rab38* is
2 selected from the group consisting of at least one *Rab38* exon, at least one *Rab38* intron, the
3 *Rab38* 5' untranslated sequence, and the *Rab38* 3' untranslated sequence.
- 1 10. The method of Claim 9, wherein said at least one *Rab38* exon is
2 selected from the group consisting of *Rab38* exon 1, *Rab38* exon 2, and *Rab38* exon 3.
- 1 11. The method of Claim 7, wherein said genomic DNA is mammalian
2 genomic DNA.
- 1 12. The method of Claim 7, wherein said purifying is accomplished using
2 size selection.

1 13. A method for detecting mutations in *Rab38* comprising the steps of:
2 a) amplifying at least a portion of *Rab38* from genomic DNA to
3 yield a *Rab38* amplification product;
4 b) digesting said *Rab38* amplification product to yield a digested
5 *Rab38* amplification product; and
6 c) electrophoresing said digested *Rab38* amplification product.

1 14. The method of Claim 13, wherein said amplifying is accomplished
2 using a polymerase chain reaction.

1 15. The method of Claim 13, wherein said at least a portion of *Rab38* is
2 selected from the group consisting of at least one *Rab38* exon, at least one *Rab38* intron, the
3 *Rab38* 5' untranslated sequence, and the *Rab38* 3' untranslated sequence.

1 16. The method of Claim 15, wherein said at least one *Rab38* exon is
2 selected from the group consisting of *Rab38* exon 1, *Rab38* exon 2, and *Rab38* exon 3.

1 17. The method of Claim 13, wherein said genomic DNA is mammalian
2 genomic DNA.

1 18. A method for screening for biologically active agents to modulate
2 RAB38 activity, comprising the steps of:

3 a) providing:
4 i) melanocytes comprising RAB38 activity, and
5 ii) a candidate agent; and
6 b) exposing said melanocytes to said candidate agent to yield
7 treated melanocytes; and
8 c) measuring the modulation of said RAB38 activity of said
9 treated melanocytes by said candidate agent.

1 19. The method of Claim 18, wherein said RAB38 activity comprises
2 GTPase activity.

1 20. The method of Claim 18, wherein said RAB38 activity comprises GTP
2 binding activity.

1 21. The method of Claim 18, wherein said RAB38 activity comprises GDP
2 release.

1 22. The method of Claim 18, wherein said RAB38 activity comprises
2 TYRP1 trafficking to melanosomes.

1 23. The method of Claim 18, wherein said RAB38 activity comprises
2 RAB38 trafficking to melanosomes.

1 24. A kit for screening for biologically active agents that modulate RAB38
2 activity, comprising: a) plurality of melanocytes comprising RAB38 activity, wherein said
3 melanocytes are provided within a container, and b) instructions for determination of RAB38
4 activity in said melanocytes.

1 25. The kit of Claim 24, further comprising means to analyze RAB38
2 activity.

1 26. The kit of Claim 25, wherein said means to analyze RAB38 activity
2 comprises an assay to assess GTPase activity.

1 27. The kit of Claim 25, wherein said means to analyze RAB38 activity
2 comprises an assay to assess GTP binding activity.

1 28. The kit of Claim 25, wherein said means to analyze RAB38 activity
2 comprises an assay to assess GDP release.

1 29. The kit of Claim 25, wherein said means to analyze RAB38 activity
2 comprises an assay to assess TYRP 1 trafficking to melanosomes.

1 30. The kit of Claim 25, wherein said means to analyze RAB38 activity
2 comprises an assay to assess RAB38 trafficking to melanosomes.

1 31. A kit for detection of mutations in RAB38 comprising at least two
2 primer sequences suitable for amplification of at least a portion of RAB38, and instructions
3 for utilizing said kit.

1 32. The kit of Claim 31, wherein said primer sequences are selected from
2 the group consisting of SEQ ID NO:17, SEQ ID NO:18, SEQ ID NO:19, SEQ ID NO:20,
3 SEQ ID NO:21, and SEQ ID NO:22.

1 33. The kit of Claim 31, wherein said kit is suitable for use in the
2 polymerase chain reaction.

1 34. The kit of Claim 31, further comprising reagents for digesting nucleic
2 acid.

1 35. A kit for diagnosing defects in melanosome function, comprising
2 melanocytes comprising *RAB38* and instructions for assessing defects in melanosome
3 function.

1 36. The kit of Claim 35, further comprising means to analyze *RAB38*
2 activity.

1 37. The kit of Claim 36, wherein said means to analyze *RAB38* activity
2 comprises an assay to assess GTPase activity.

1 38. The kit of Claim 36, wherein said means to analyze *RAB38* activity
2 comprises an assay to assess GTP binding activity.

1 39. The kit of Claim 36, wherein said means to analyze *RAB38* activity
2 comprises an assay to assess GDP release.

1 40. The kit of Claim 36, wherein said means to analyze *RAB38* activity
2 comprises an assay to assess TYRP1 trafficking to melanosomes.

1 41. The kit of Claim 36, wherein said means to analyze *RAB38* activity
2 comprises an assay to assess *RAB38* trafficking to melanosomes.

1 42. A composition for modulating pigmentation of melanocytes,
2 comprising a modulator of *RAB38* activity.

1 43. The composition of claim 42, wherein the modulator of *RAB38*
2 activity is an enhancer of *RAB38* activity.

1 44. The composition of claim 42, wherein the modulator of RAB38
2 activity is an inhibitor of RAB38 activity.

1 45. The composition of claim 44, wherein the inhibitor of RAB38 activity
2 is selected from the group consisting of siRNA and intrabodies.

1 46. A method of modulating the pigmentation of a melanosome and
2 changing skin color, the method comprising: contacting a skin surface with a modulator of
3 RAB38 activity, thereby regulating the activity of RAB38.

1 47. The method of claim 46, wherein the modulator is an inhibitor of
2 RAB38 activity that down-regulates RAB38 activity and lightens skin color.

1 48. The method of claim 46, wherein the modulator is an enhancer of
2 RAB38 activity that up-regulates RAB38 activity and darkens skin color.